Usher Syndrome Type 2C

National Cancer Institute

Source

National Cancer Institute. Usher Syndrome Type 2C. NCI Thesaurus. Code C153174.

An autosomal recessive sub-type of Usher syndrome caused by homozygous or compound heterozygous mutation(s) in the ADGRV1 gene, encoding adhesion G protein-coupled receptor V1. It may also result from biallelic digenic mutation(s) in ADGRV1 and PDZD7, which encodes PDZ domain-containing protein 7.